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CASE REPORT

Reversal of haemochromatotic cardiomyopathy in β thalassaemia by chelation therapy

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Abstract

Haemochromatotic cardiomyopathy is the main cause of morbidity and mortality in patients with β thalassaemia major. Once congestive heart failure develops most patients die in a few months.

Congestive heart failure was reversed and echocardiographic findings were restored to normal in a 24 year old woman with β thalassaemia who resumed treatment with chelation therapy (desferrioxamine).

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Keywords: haemochromatotic cardiomyopathy, β thalassaemia, chelation therapy.

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In patients with the severe transfusion-dependent form of β thalassaemia large quantities of iron accumulate and produce a clinical syndrome resembling idiopathic haemochromatosis, with liver, heart, and endocrine dysfunction.[1] During the past decade management of β thalassaemia has been improved by new transfusion regimens and a more effective iron chelation therapy with subcutaneous desferrioxamine (DFX).[1 2] Though these measures have substantially changed the course of the illness, cardiac dysfunction remains the primary cause of morbidity and mortality.[1 2] Once congestive heart failure develops, most patients die in a few months, generally in the second or third decade of life.[1]

We report a case in which cardiomyopathy with congestive heart failure was reversed by resumption of chelation therapy with DFX.

Case report

A 24 year old splenectomised woman with β thalassaemia major had been treated with regular transfusions and subcutaneous DFX since the age of two. Four years before she stopped coming regularly for DFX treatment and her serum ferritin concentration increased. During the next 2 years diabetes mellitus that required insulin treatment developed and a few months later she complained of increasing breathlessness and showed evidence of congestive heart failure. Clinical examination showed tachycardia, hypotension, rales, gallop rhythm, considerable liver enlargement, pleural effusion on the chest *x* ray, and inverted T waves on the electrocardiogram. Echocardiography showed greatly increased left ventricular end diastolic (61 mm) and end systolic (56 mm) cavity dimensions (fig 1) and reduced percentage of fractional shortening (8%). The left ventricular ejection fraction was 25% and there was Doppler evidence of moderate mitral regurgitation and mild tricuspid regurgitation, with an estimated systolic pulmonary pressure of 40 mm Hg. The serum ferritin concentration was 5150 ng/ml.

Treatment with digoxin and diuretics was started and treatment with subcutaneous DFX was resumed (50 mg/kg body weight daily). Despite a gradual improvement in the congestive heart failure echocardiographic evidence of dilated cardiomyopathy persisted.

Over more than a year the left ventricular cavity dimension and ejection fraction gradually improved and the serum ferritin concentration decreased to 1200 ng/ml. Digoxin and diuretics were withdrawn after 16 months. Two years after the resumption of DFX treatment the patient was symptom free and the

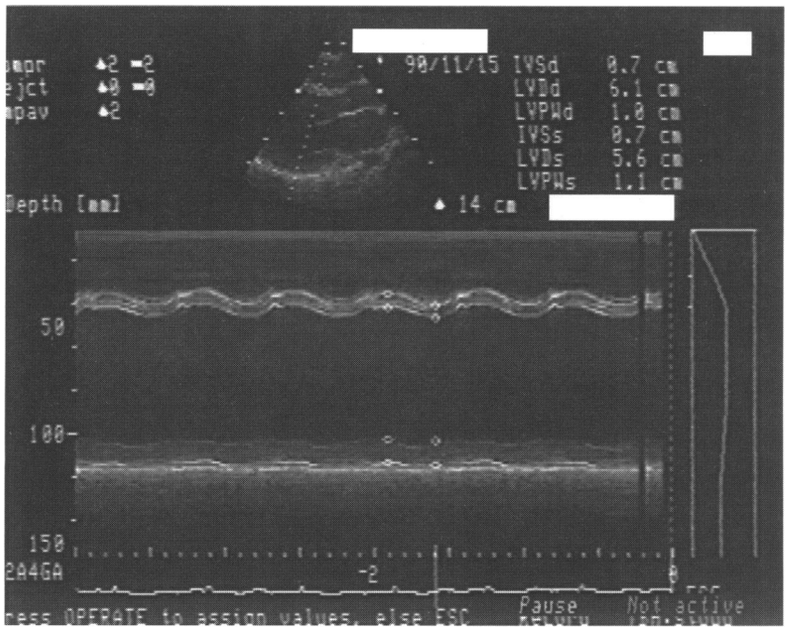

Figure 1 M mode echocardiogram showing increase of left ventricular end diastolic (61 mm) and end systolic (56 mm) cavity dimension and reduced percentage of fractional shortening (8%).

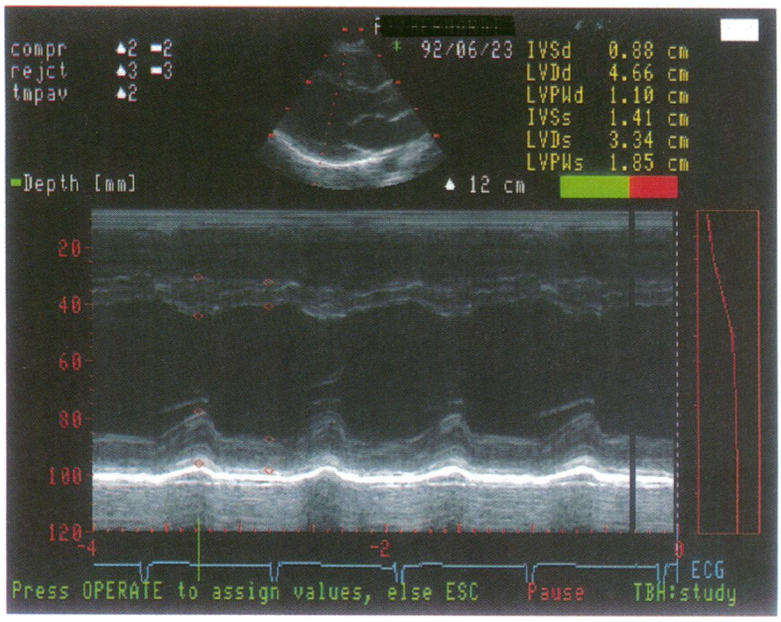

Figure 2 M mode echocardiogram showing normal left ventricular end diastolic (46 mm) and end systolic (33 mm) cavity dimension and percentage fractional shortening (28%) after chelation therapy.

echocardiogram was normal (fig 2). She continues to be treated regularly with DFX.

Discussion

The cardiac complications of β thalassaemia major include pericarditis, arrhythmias, right heart failure with pulmonary hypertension,[3] high cardiac output, and restrictive[4] and dilated cardiomyopathy, which is the most common cause of death.[1 2]

Our patient might have recovered spontaneously from myocarditis. However, the clinical, laboratory, and echocardiographic findings indicate that the most probable explanation is that dilated cardiomyopathy was caused by iron deposition which was reversed by resumption of chelation therapy. This is consistent with recent studies showing that compliance with subcutaneous DFX is important in preventing[1] and reversing (by intensified therapy)[2] cardiac dysfunction in β thalassaemia major and with an earlier report that congestive cardiomyopathy caused by iron overload in a patient with β thalassaemia intermedia[5] and a patient with megaloblastic anaemia[6] was reversed by chelation therapy.

Improved management of the cardiac complications of β thalassaemia major includes detection and treatment of arrhythmias and treatment with vasodilators and inotropic agents as well as diuretics and digoxin in case of heart failure.

This case indicates that patients at risk of iron overload must be encouraged to comply with chelation therapy.

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